National Experts Program
برنامج خبراء الإمارات

AI Report

Healthy Haven – A World Where Children Thrive, Enabled by Artificial Intelligence and Genetic Sequencing

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Healthcare and Well-Being

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Fatima Al-Ali has 11 years of experience in healthcare management and operations. She holds a Bachelor of Science in Business Administration degree from the American University of Sharjah, with a major in Finance.

Al-Ali is currently managing multiple transformation and expansion projects across Mubadala Health's network of assets. She previously served as the CEO of Abu Dhabi Telemedicine Centre where she drove digital healthcare innovation, which includes successfully introducing the first-of-its-kind primary care gatekeeping model through telemedicine. Al-Ali was also responsible for the overall administration, strategic planning, and development of the Centre.

Prior to that, she was the Operations Manager of Abu Dhabi Telemedicine Centre, responsible for overseeing operational improvements, supervising the workforce, and supporting business development efforts. She also spent three years at Mubadala Investment Company managing operations of its Wooridul Spine Centre as well as working on strategy, business development and finance projects for Cleveland Clinic Abu Dhabi.

Advances in technology and the digitization of services are driving greater need for personalization in industries including healthcare. The relationship between healthcare providers and patients is predicted to transform radically, with genomics and artificial intelligence (AI) enabling care that is more patient-centered and personalized.

The process of personalizing healthcare depends heavily on understanding our genetic profile. This involves genetic sequencing, which analyzes a patient's complete DNA composition.

> **Genetic sequencing can help healthcare providers determine a patient's potential health risks—and develop tailored therapies and treatment plans specific to that patient's genetic makeup.**

It has the power to exponentially improve the accuracy of disease diagnosis.

Genetic sequencing can also work more effectively in the treatment of rare diseases for which conventional treatments have failed.

The basics

To understand how AI and genetic sequencing can change the future of medicine, we need to understand the basics of genomics.

Genomics England, a British organization that specializes in genomic healthcare, describes a genome as the complete set of genetic instructions that determine the traits (characteristics and conditions) of an organism .[1] It is made up of DNA, genes, and chromosomes.

There are two types of genome sequencing: whole-genome sequencing (WGS) and targeted genomic sequencing (TS):

WGS involves analyzing the entire genome of an organism—and is a powerful tool for research and sequencing studies. It provides a comprehensive view of an organism's genetic makeup, including regions of the genome that may have not been studied, and information on genetic variations that are directly linked to defects and diseases.

TS, on the other hand, focuses on a specific component of the genome and is often conducted to gain more insight on a particular disease or trait.

Genetic mutations are a type of genetic variation that occur when cell division causes a change in the DNA sequence. The severity and impact of genetic mutations varies depending on where they occur and what change they cause. Some mutations are beneficial, such as abnormally high bone density. But other mutations are harmful and can lead to genetic disorders like hemophilia and cancers.[2]

Converting genomic data to action through AI

There is a huge volume of genome data. Translating large and sophisticated genomic datasets into actionable clinical information is critical to the delivery of personalized medicine - but requires computer automation.

> **AI will play an important role in accelerating the analysis of a population's complex genomic data.**

This can be achieved through machine learning and deep learning in three key areas:

Predictive Modeling: Identifying patterns in data and better understanding how genetic variants are linked with specific diseases or traits

Clustering and Classification: Grouping individuals with genetic similarities or differences

Causal Analysis: Identifying the causes of specific diseases in the measured data and using cause-and-effect analysis to develop personalized treatment

UAE launches pioneering human genomics initiative

In the UAE, the Department of Health – Abu Dhabi has launched a pioneering human genomics initiative it describes as the world's most comprehensive genome program. The Emirati Genome Program (EGP) is a national project that will use large-scale genomic data "to improve health and well-being".[3]

The Department of Health says the EGP will advance the nation's personalized healthcare agenda. It aims to be the first of its kind worldwide to provide citizens with their own high-quality genome as standard—and to incorporate genomic data into healthcare management. This will be achieved by using advanced sequencing technology to generate the highest quality, most comprehensive genome data.

1 What Are Genes, DNA, and Chromosomes? verywellhealth.com, 30 October 2022
2 Genetic Mutations in Humans, Cleveland Clinic
3 Department Of Health unveils world's most comprehensive Genome Program, Transforming health and well-being with genomics and Artificial Intelligence, the nation's leading strengths, UAE Department of Health, 10 December 2019

Enabled through AI, programs like these provide countries with opportunities to better predict future diseases and improve public health initiatives. They provide a foundation for further research and improve cost management through targeted therapies. A number of clinical programs have been piloted successfully by leveraging the EGP data to implement personalized and targeted therapies. These have included programs to reduce the burden of breast cancer in women and the burden of hereditary diseases in children.

The rising trend of consanguineous marriages poses a serious threat to future generations of being born with genetic disorders

Consanguineous marriages—those between blood relatives—are common in Middle Eastern societies, including the UAE. Such relationships are linked to an increased risk of genetic disorders and birth defects in children, so the rising trend of these types of marriages poses a serious threat to future generations. About **75** of every **1,000 babies** in the UAE are born with a birth defect, according to data from Dubai's Centre for Arab Genomic Studies [4]. This ranks the UAE sixth out of **193 countries** for children born with birth defects. Separate research published in the journal Clinical Genetics found children born to consanguineous unions also had significantly higher incidences of illnesses (**37.1%**) than those of non-consanguineous unions (**29%**) [5].The occurrence of malignancies, congenital abnormalities, intellectual disability, and physical disability was also significantly higher in offspring of consanguineous marriages. This has led to the introduction of compulsory premarital screening for all couples that includes tests for infectious diseases and a limited number of genetic blood disorders.

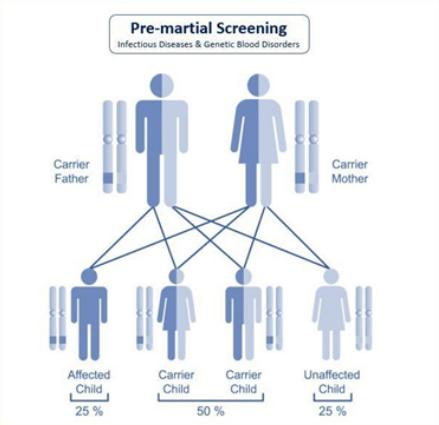

Exhibit 1

Current premarital screenings include a very limited number of genetic tests, which may not be effective in identifying carrier couples of serious genetic disorders. If both parents are carriers, the probability of having an affected child is 25% at each pregnancy.

In November 2022, the Department of Health – Abu Dhabi announced the incorporation of an optional genetic screening in its premarital screening program. This covers tests for **570 genes** [6] causing autosomal recessive disorders—those disorders inherited from both parents. This optional genetic screening will be supported by the Emirati Genome Program and will deploy AI through risk stratification techniques. For new couples, the optional screening will help to identify the causative genes for diseases their children may carry. His Excellency Dr. Omniyat Al Hajeri, Executive Director of the Community Health Sector in Abu Dhabi Public Health Center, said the screening initiative would "contribute to building a healthier and disease-free society with less treatment burden on the parents and on the healthcare sector." [7]

Positive disease carrier couples will be referred to genetic counseling to discuss their carrier status. Alternative options for conception, such as in-vitro fertilization (IVF), can also be considered.

IVF can help reduce the transmission of genetic diseases to a couple's offspring by using a technique called preimplantation genetic testing (PGT). PGT involves the screening of embryos for genetic mutations before they are implanted to ensure that only embryos without the disease-causing genes are implanted. This reduces the risk of giving birth to children with serious genetic disorders. PGT is not a foolproof method, as there are limitations to what it can detect, but the combination of IVF and PGT can significantly improve the chances of giving birth to a healthy child.

Over time, these alternative conception efforts can gradually eliminate harmful genetic variations from the population, generation by generation. This could ultimately lead to a future where inherited diseases are much less common—or even eradicated entirely.

4 Science offers answers to family marriage gene problems, thenationalnews.com, 17 November 2012
5 Abdulrazzaq YM, Bener A, al-Gazali LI, al-Khayat AI, Micallef R, Gaber T. A study of possible deleterious effects of consanguinity, Clinical Genetics, 28 June 2008

6 DoH to expand pre-marital screening for 570 genetic medical conditions, Department of Health, doh.gov.ae, 25 November 2022

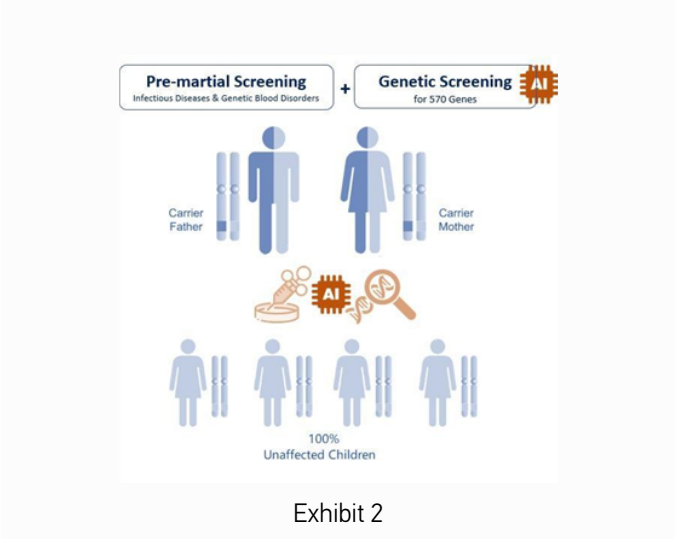

Exhibit 2

Incorporating an expanded list of genetic tests into premarital screening, combined with IVF for positive disease carrier couples, significantly increases the chance of healthy children.

The role of AI in IVF

Many factors determine the success of IVF cycles. These include the patient's age, medical history, and gamete quality—the quality of the male and female reproductive cells.

> **Research suggests AI has great potential to revolutionize IVF treatment, from predicting success rates to developing personalized treatment regimes.**

01 Gamete and embryo selection:
The IVF process relies heavily on the experience of embryologists to manually assess gamete health and grade embryos to choose the best one for implantation. A light microscope or time-lapse imaging is used to visually assess reproductive cells and embryos. By using machine learning to read and assess images, AI can provide a more objective and accurate assessment of gametes and ranking of embryos, according to research led by The University of Adelaide in Australia. [7]

02 Predicting success rates:

> **By analyzing a patient's medical history and previous IVF outcomes, AI can help doctors predict the chance of success of IVF treatment.**

This helps doctors make informed decisions, such as whether to proceed with IVF or to try other treatments. IVF clinics can use AI algorithms to analyze large datasets of patients and treatment data to identify patterns and correlations that can improve success rates. This can help clinics identify and address underlying factors that contribute to the success of IVF procedures.

03 Personalized treatment regimens:
IVF stimulation protocols and treatment regimens differ from one patient to another. Physicians determine treatment regimens based on their clinical experience or research. AI can support the decision-making process and analyze hormonal stimulation protocols used in IVF to suggest personalized treatment regimens for each patient.

04 Advanced imaging and diagnostics:
For women undergoing reproductive treatments, multiple examinations are needed to monitor the development and growth of follicles throughout each IVF cycle. This is time-consuming for clinicians and inconvenient for patients. AI-aided ultrasound techniques in follicular monitoring are needed to increase accuracy, optimize images, and improve the use of the clinician's and patient's time.

Next steps—expanding these initiatives to maximize impact

01 Intervene at the pre-conception stage:
The preparation stage before IVF starts is vitally important and typically involves several steps that are designed to optimize outcomes. These include:

I. Comprehensive medical evaluations to determine any factors affecting fertility, such as ovarian function and hormone levels
II. Making lifestyle changes and dietary adjustments to optimize fertility
III. Taking medication to regulate hormone levels or increase the quality and number of follicles in the ovaries before the IVF cycle

Intervention at this stage can have a significant impact on the success of treatment. As described above, AI and machine learning can further improve outcomes at this stage. Leveraging specialist expert knowledge and analyzing data from existing hospital patient records can help us further understand what causes the diseases that can affect children of positive couples. AI can also support better lifestyle and health monitoring

7 Chow DJX, Wijesinghe P, Dholakia K, Dunning KR, Does artificial intelligence have a role in the IVF clinic? Reproduction and Fertility, 15 September 2021.

by capturing health data specific to each patient. This can then be analyzed and shared with clinicians to support their decision-making process.

02 Expand pilot initiatives, standardize tests, and mandate them for consanguineous couples:
Implementing these pilot initiatives at a national scale and standardizing the list of tests across all healthcare providers will eliminate variability and deliver greater benefits to society. Moving from voluntary testing to mandatory testing for confirmed consanguineous couples is also now recommended, given the high health risks in offspring associated with consanguineous marriages.

03 Provide a comprehensive reimbursement scheme for positive disease carrier couples:
The cost of treating these disorders inherited from both parents is significantly higher than the cost of an IVF cycle. A very common autosomal disorder is cystic fibrosis, an inherited disease that causes severe damage to the lungs and requires more intensive treatment as it progresses. The lifetime healthcare costs of cystic fibrosis have been estimated as **$306,332**.[8] This is **10 times** higher than a single IVF cycle.

The economic impact of treating these disorders goes beyond the healthcare-associated costs. According to researchers at the University of Hong Kong, rare diseases are often chronically debilitating and need lifelong care by paid formal caregivers or informal caregivers.[9] These are typically family members or friends providing non-professional and unpaid care and support. In other words, indirect societal costs beyond those of the healthcare system must be considered in a comprehensive evaluation of the socio-economic impact of rare diseases.

A single IVF cycle, from start to end, can range from **$15,000** to **$30,000**, yet most insurance schemes do not cover fertility treatments—let alone for couples who may not have fertility issues to begin with. It is critical, then, that we address this gap by providing an appropriate reimbursement structure to cover IVF treatments for disease carrier couples. By rolling out AI applications, healthcare costs can be allocated effectively, and the impact of reimbursement models maximized.

04 Conduct a qualitative study to understand the reproductive and genetic consequences of consanguinity in live marriages:
A qualitative study will help us better understand the impact of consanguinity in marriage. Data points to consider in this study may include the number of live births, the incidence of illness in offspring, the most common illnesses in offspring, the types of genetic mutations in parents, and the level of consanguinity between parents (first, second, or third cousins).

A qualitative study can also identify valuable correlations between the data collected—for example, the correlation between genetic mutations in parents and illnesses in their offspring. Study results can also help researchers and clinicians identify alternative interventions to support parents in having healthy children in future pregnancies.

8 van Gool K, Norman R, Delatycki MB, Hall J, Massie J, Understanding the costs of care for cystic fibrosis: an analysis by age and health state, Value in Health, March 2013
9 Claudia C.Y. Chung, Nicole Y.T. Ng, Yvette N.C. Ng, Adrian C.Y. Lui, Jasmine L.F. Fung, Marcus C.Y. Chan et al, Socio-economic costs of rare diseases and the risk of financial hardship: a cross-sectional study, The Lancet, 22 February 2023